



Genetic population analysis of 17 Y-chromosomal STRs in three states (Valle del Cauca, Cauca and Nariño) from Southwestern Colombia

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ARTICLE INFO

Article history:

Received 9 November 2007

Received in revised form 4 September 2008

Accepted 9 December 2008

Available online 17 January 2009

Keywords:

Forensic science

Y-Chromosome

DNA typing

STR

Haplotype

DYS19

DYS385a

DYS385b

DYS389I

DYS389II

DYS390

DYS391

DYS392

DYS393

DYS438

DYS439

DYS437

DYS448

DYS456

DYS458

DYS635

GATA-H4

Colombia

Population genetics

ABSTRACT

Seventeen Y-chromosomal (DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS438, DYS439, DYS437, DYS448, DYS456, DYS458, DYS635, YGATA-H4 and DYS385a/b) short tandem repeat (STR) polymorphic systems were typed in three South West Colombian populations: Valle (short term for Valle del Cauca), Cauca and Nariño. DYS385a/b showed the highest gene diversity in the three populations. A total of 287 different Y-chromosome haplotypes were observed in the 308 males analyzed, and the haplotype diversity among populations was 0.9977. The most frequent haplotype was observed only three times and only nineteen others were observed two times. The highest gene diversity was found in Valle and the lowest in Cauca. Analysis of molecular variance (AMOVA) revealed that variation is mainly within populations (99.1%) in agreement with previous results in European populations. In conclusion, these populations could be pooled together in order to create one "Colombian-Mestizo" database for forensic use.

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1. Introduction

The non-recombinant Y-chromosome segment shows a paternally inherited haploid transmission pattern. Because Y-STRs can be employed to construct highly discriminative Y-haplotypes, they are useful in stain analysis, paternity testing and other forensic genetic studies on the basis of their male-specificity. From the evolutionary point of view, these markers are useful for tracing migration movements of populations and inferring the history of

modern humans. These markers allow amplification of male-specific DNA regions in a male/female mixture without female DNA competition for reagents during PCR. The gene frequencies and gene and haplotype diversities for 17 Y-STR loci reported here in three Southwestern states from Colombia will contribute to the world database and to the knowledge on genetic polymorphisms of local populations.

2. Methods

Blood samples were obtained from 308 reportedly unrelated healthy donors born in the Southwestern Colombian states of

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Table 1
Y-chromosome STR haplotypes in three Colombian populations.

Haplotype	DYS19	DYS385a/ b	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS438	DYS439	DYS437	DYS448	DYS448	DYS456	DYS458	DYS635	GATA H4	VALLE	CAUCA	NARIÑO	Total: 308
1	14	14-11	13	29	24	10	13	13	12	11	15	19		16	16	24	12	1			1
2	16	16-11	13	30	25	11	11	13	11	10	14	20		15	15	23	13	1			1
3	14	14-13	13	30	23	10	11	12	10	11	14	21		15	17.2	21	11	1			1
4	13	13-14	13	31	25	10	13	13	11	11	15	19		16	17	23	11	1			1
5	14	14-11	13	29	24	11	13	13	11	12	14	18		15	18	23	10	1			1
6	14	14-14	12	28	25	11	11	13	11	12	14	19		15	18	24	11	1			1
7	13	13-16	12	29	25	10	11	12	10	12	14	21		15	19	21	11	1			1
8	13	13-13	14	30	24	9	11	13	10	10	14	20		16	19	21	12	1			1
9	13	13-16	13	31	23	10	11	13	10	12	14	21		15	20	21	11	1			1
10	14	14-11	12	28	24	11	13	13	12	11	15	19		15	18	23	12	1			1
11	14	14-13	13	31	23	10	11	12	9	9	14	20		15	16.2	19	11	1			1
12	14	14-11	14	30	26	11	13	8	12	12	15	19		12	16	23	13	1			1
13	14	14-11	13	29	24	10	13	13	12	12	15	19		16	17	23	12	2			2
14	14	14-15	12	29	24	6	14	13	11	11	14	19		15	16	23	11	1			1
15	14	14-10	13	29	24	11	13	13	12	13	15	19		16	21	23	12	1			1
16	14	14-11	13	29	25	11	13	13	12	12	15	19		16	17	23	12	1			2
17	14	14-12	14	29	22	10	11	12	9	11	15	22		15	19	22	11	1	1		2
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19	14	14-13	12	28	22	10	11	13	10	12	16	20		15	15	22	11	1			1
20	14	14-13	14	31	24	10	11	12	9	11	15	20		17	18	25	12	1			1
21	13	13-15	13	29	23	6	14	11	11	11	14	19		16	15	22	11	1			1
22	14	14-12	13	29	24	11	13	13	12	12	15	19		16	17	24	11	1			1
23	15	15-11	13	31	24	12	11	13	11	10	14	20		15	18	23	13	1			1
24	14	14-11	13	29	26	11	13	13	12	12	15	19		16	17	23	12	1			1
25	14	14-13	13	30	23	10	11	12	9	11	15	20		16	18	24	11	1			1
26	14	14-11	14	30	24	10	13	13	12	11	15	19		15	17	23	12	1			1
27	14	14-11	13	28	23	11	12	13	12	11	15	19		15	18	23	11	1			1
28	14	14-11	13	29	24	10	13	13	13	13	14	18		17	16	23	11	1			1
29	14	14-11	13	29	26	11	13	12	12	12	15	19		15	16	24	11	1			1
30	14	14-11	14	30	24	9	13	13	12	12	14	18		16	17	23	11	1			1
31	15	15-12	12	29	24	10	11	14	10	11	14	20		15	16	21	11	1			1
32	14	14-11	12	28	24	11	13	13	12	12	15	19		16	16	23	12	1	1		2
33	15	15-16	13	30	22	10	11	14	11	11	16	20		16	17	22	11	1			1
34	14	14-11	13	29	24	11	13	13	12	12	15	19		16	19	23	12	1			1
35	15	15-16	13	30	22	10	11	13	10	12	15	21		15	16	22	11	1			1
36	15	15-11	13	29	23	10	13	13	12	12	15	20		16	18	23	12	1			1
37	13	13-13	12	30	25	11	14	13	11	12	14	18		18	18	22	11	1			1
38	14	14-13	12	28	24	10	11	13	10	11	16	20		14	15	21	11	1			1
39	14	14-11	13	29	24	11	13	13	12	12	14	18		16	17	23	12	1			1
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46	14	14-11	13	29	24	11	13	13	12	11	15	20		15	17	23	11	1			1
47	15	15-13	13	29	23	9	11	13	9	12	14	20		17	15	22	12	1			1
48	14	14-11	13	29	24	11	13	13	12	13	15	19		18	17	23	12	1			1
49	14	14-11	13	29	25	10	13	12	12	11	15	19		15	15	23	12	1			1
50	14	14-14	13	30	24	11	13	13	13	11	15	20		16	16	23	12	2			2
51	13	13-14	13	29	22	10	16	13	11	11	14	19		15	17	22	10	1			1
52	14	14-12	13	29	24	11	13	13	12	14	15	19		16	17	23	12	1			1
53	16	16-14	12	28	24	10	11	12	9	11	15	19		13	16	21	11	1			1
54	15	15-11	13	30	23	11	11	13	11	10	14	20		16	17	23	12	1			1
55	14	14-11	13	29	24	11	13	13	12	12	15	19		16	17	23	12	1			1

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Table 1 (continued)

Haplotype	DYS19	DYS385a/ b	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS438	DYS439	DYS437	DYS448	DYS448	DYS456	DYS458	DYS635	GATA H4	VALLE	CAUCA	NARIÑO	Total: 308
56	14	14-11	13	29	25	10	13	13	12	12	15	18		16	20	23	12	1			1
57	14	14-13	14	32	23	10	11	12	10	11	14	20		15	17.2	21	11	1	2		3
58	15	15-13	12	27	25	10	14	13	9	11	14	19		15	19	21	11	1			1
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60	15	15-11	13	29	24	11	13	13	12	11	15	19		15	16	23	12	1			1
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66	13	13-14	14	33	23	11	11	12	9	10	15	19		16	14	22	11	1			1
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68	15	15-17	12	29	24	10	11	13	10	12	14	20		17	15	20	13	1			1
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115	13	13-14	13	30	23	10	15	13	11	11	14	19	14	17	25	12	1	1
116	14	14-11	14	30	24	11	13	13	12	11	15	19	15	17	23	12	1	1
117	13	13-14	12	29	23	10	14	13	11	13	13	20	17	17	24	12	2	2
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123	14	14-11	14	30	24	10	13	14	12	12	15	18	17	18	23	12	1	1
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141	13	13-13	14	30	24	9	11	13	10	10	14	20	17	19	21	12	1	1
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143	14	14-11	13	29	24	10	12	13	12	13	15	19	16	17	23	12	1	1
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160	15	15-15	13	31	21	12	11	13	11	12	14	21	15	18	22	11	1	1
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165	13	13-14	13	30	25	10	14	13	11	11	14	19	15	17	22	11	1	1
166	15	15-16	12	28	24	10	11	12	9	12	14	20	14	18	20	9	1	1
167	17	17-11	13	28	23	10	11	13	10	11	15	22	14	17	23	14	1	1
168	17	17-12	13	28	23	10	11	13	10	11	15	21	14	17	22	13	2	2
169	14	14-11	13	29	24	11	13	13	12	11	15	19	16	16	23	12	1	1
170	13	13-14	13	31	23	10	14	13	11	11	14	19	15	18	22	11	1	1
171	14	14-11	13	29	24	10	13	13	12	12	15	19	15	17	23	12	2	2

(continued on next page)

Table 1 (continued)

Haplotype	DYS19	DYS385a/ b	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS438	DYS439	DYS437	DYS448	DYS448	DYS456	DYS458	DYS635	GATA H4	VALLE	CAUCA	NARIÑO	Total: 308
172	13	13-15	12	30	23	10	15	13	13	11	14	19		15	15	22	12		1		1
173	15	15-14	12	29	23	9	11	12	9	12	14	21		16	14	22	12		1		1
174	13	13-14	13	31	23	10	14	13	11	11	14	19		10	16	22	12		1		1
175	14	14-12	13	29	23	11	13	12	12	11	15	19		15	17	23	12		1		1
176	13	13-14	14	31	24	10	14	13	11	12	15	19		15	17	22	12		1		1
177	14	14-11	13	29	24	11	13	13	12	11	15	19		16	16	23	11		1		1
178	13	13-16	13	31	24	9	11	14	10	13	14	20		15	17	24	11		1		1
179	13	13-15	14	30	24	10	15	13	11	12	14	20		15	16	23	12		2		2
180	14	14-11	13	29	23	11	13	13	12	11	15	19		15	17	23	11		1		1
181	13	13-15	13	30	24	10	15	13	12	12	14	20		15	16	23	12		1		1
182	14	14-13	13	29	23	10	11	12	10	13	14	20		15	19.2	21	11		1		1
183	17	17-12	14	29	23	10	11	13	10	11	15	20		14	16	22	12		1		1
184	16	16-12	14	31	24	10	11	14	10	10	14	20		15	16	21	11		1		1
185	14	14-11	13	29	23	11	13	12	12	15	19			16	17	23	12		1		1
186	14	14-13	14	30	23	10	11	12	10	10	14	20		15	17.2	20	11		1		1
187	15	15-16	14	31	21	10	11	13	11	12	14	21		15	17	22	10		1		1
188	15	15-11	14	32	23	11	13	13	12	11	14	19		16	17	23	12		1		1
189	15	15-16	12	28	24	10	11	12	9	12	14	20		14	18	21	9		1		1
190	13	13-15	13	32	25	10	15	13	13	11	14	19		14	15	22	12		1		1
191	13	13-13	14	30	24	9	11	13	11	10	14	20		16	18	21	13		1		1
192	17	17-12	13	28	23	10	11	13	10	11	15	20		14	16	22	12		1		1
193	15	15-12	12	29	21	10	11	15	10	11	16	21		15	16	22	11			1	1
194	14	14-14	13	29	23	11	13	13	12	12	15	19		16	17	23	12			1	1
195	14	14-11	13	30	26	11	13	13	12	12	15	19		15	17	23	11			1	1
196	14	14-11	14	30	25	10	13	13	11	13	15	19		15	16	23	12			1	1
197	13	13-13	14	32	24	11	16	13	11	12	14	20		15	16	22	12			1	1
198	13	13-13	13	29	24	9	11	13	10	10	14	20		17	18	21	12			1	1
199	17	17-13	12	30	22	10	12	12	10	10	17	19	20	15	15	20	11			1	1
200	15	15-11	13	29	24	11	13	13	12	11	15	18		17	16	23	12			1	1
201	14	14-11	13	29	24	10	13	12	11	12	15	20		16	16	23	12			1	1
202	13	13-17	13	30	24	11	11	13	10	11	14	20		18	16	22	11			2	2
203	13	13-13	14	32	24	11	14	13	11	12	14	20		15	16	22	12			1	1
204	14	14-11	14	30	24	10	13	13	12	10	14	18		15	17	23	11			1	1
205	15	15-11	13	29	24	11	13	13	12	12	15	18		17	16	23	12			1	1
206	13	13-15	12	31	23	10	15	13	13	11	14	19		15	15	22	13			1	1
207	13	13-15	12	30	23	10	15	12	12	11	14	19		15	15	22	12			1	1
208	13	13-14	14	31	23	11	14	12	10	13	14	20		16	16	22	12			1	1
209	14	14-11	14	30	24	11	13	13	12	11	14	19		16	17	23	12			2	2
210	13	13-14	13	30	24	10	14	13	11	12	14	19		15	15	21	11			1	1
211	13	13-11	13	29	24	11	13	13	12	12	15	20		16	16	23	12			1	1
212	14	14-12	13	28	23	10	11	13	10	11	15	21		15	17	22	13			1	1
213	14	14-11	13	29	24	10	12	13	12	12	15	19		15	18	23	12			1	1
214	14	14-12	13	30	24	11	13	13	12	11	15	19		17	18	23	12			1	1
215	13	13-16	12	31	22	10	15	13	12	11	14	19		16	15	22	11			1	1
216	13	13-14	14	30	24	10	14	12	11	12	14	20		14	17	22	12			1	1
217	13	13-15	13	30	24	10	11	13	10	13	14	19		17	15	23	12			1	1
218	14	14-11	13	29	24	11	13	13	12	13	15	19		16	17	23	11			1	1
219	13	13-13	14	30	24	9	11	13	10	10	14	20		16	18	22	12			1	1
220	13	13-17	13	29	25	9	11	14	10	11	14	20		15	16	23	11			1	1
221	15	15-16	13	32	21	10	11	13	11	11	14	21		15	16	21	11			1	1
222	14	14-11	13	29	24	10	13	13	12	12	15	19		12	18	24	12			1	1
223	14	14-15	13	30	24	9	13	13	12	11	15	19		15	17	23	12			1	1
224	13	13-12	13	31	24	11	14	14	11	12	14	20		16	17	22	11			1	1
225	13	13-17	13	30	24	10	11	13	10	12	14	20		15	13	21	12			1	1
226	14	14-11	14	31	24	11	13	13	12	11	15	19		17	19	23	12			1	1
227	14	14-11	13	30	23	11	13	13	12	12	15	19		18	17	23	13			1	1
228	14	14-15	12	30	24	10	15	13	12	11	14	19		15	15	22	12			1	1

229	14	14-11	13	30	23	11	13	13	12	12	15	19	16	17	23	13	1	1
230	12	12-12	14	31	25	11	14	13	11	12	14	20	15	16	22	11	1	1
231	13	13-15	14	33	23	10	11	13	10	13	14	21	15	18	21	12	2	2
232	14	14-13	13	31	23	10	11	12	10	12	14	20	16	17.2	21	11	2	2
233	14	14-11	14	30	24	11	13	13	12	11	14	18	15	17	24	11	1	1
234	13	13-12	13	31	24	10	14	14	11	13	14	20	16	17	22	12	1	1
235	14	14-11	13	29	25	12	13	13	12	12	14	18	15	17	23	11	1	1
236	14	14-11	14	30	24	11	13	12	12	12	15	19	14	17	23	12	1	1
237	14	14-12	13	29	24	10	13	13	12	11	15	19	15	16	24	14	1	1
238	13	13-15	12	30	23	10	15	13	13	11	14	19	15	15	22	13	1	1
239	13	13-15	13	31	24	10	11	13	10	13	14	20	15	14	23	12	1	1
240	14	14-11	13	29	24	10	13	12	12	12	15	19	15	17	23	12	1	1
241	13	13-14	13	30	24	10	13	13	11	12	14	20	15	16	21	12	1	1
242	14	14-11	14	31	24	10	13	13	12	12	14	18	15	18	23	11	1	1
243	14	14-11	14	31	24	11	13	13	12	11	15	18	16	17	24	12	1	1
244	16	16-11	14	32	24	10	12	13	11	10	14	20	15	15	23	12	1	1
245	13	13-15	12	30	23	10	15	13	13	11	14	19	15	15	22	13	2	2
246	14	14-12	14	30	23	10	13	13	12	15	14	19	15	17	24	11	1	1
247	13	13-12	13	30	24	11	14	14	11	11	14	20	16	17	22	11	1	1
248	14	14-11	13	30	23	11	13	13	12	12	15	19	15	18	23	12	1	1
249	14	14-11	13	29	24	10	12	13	12	12	15	19	16	17	23	12	1	1
250	15	15-12	12	29	22	10	11	13	10	11	16	21	16	17	21	12	1	1
251	12	12-13	14	30	25	11	14	14	11	11	14	19	15	16	22	11	1	1
252	13	13-13	14	30	24	10	11	13	10	10	14	20	16	18	21	12	2	2
253	13	13-18	13	29	25	9	12	13	10	12	14	20	15	15	21	12	1	1
254	13	13-15	12	30	20	10	15	13	13	11	14	19	14	15	22	12	1	1
255	14	14-13	14	32	23	11	12	12	9	10	15	22	15	17	23	11	1	1
256	15	15-11	13	29	23	11	13	13	12	12	14	18	16	18	23	12	1	1
257	16	16-15	13	29	23	11	12	14	10	11	15	20	16	15	20	11	1	1
258	13	13-13	14	30	24	9	11	13	10	10	15	20	16	17	21	13	1	1
259	14	14-11	14	30	24	10	13	14	12	12	15	19	16	17	23	12	1	1
260	14	14-15	12	30	23	10	15	13	12	12	14	19	16	15	23	13	1	1
261	14	14-14	13	31	23	10	14	13	11	12	14	20	15	17	22	11	1	1
262	14	14-11	12	28	24	11	13	13	12	12	15	19	17	16	23	12	1	1
263	17	17-12	15	30	24	9	11	13	10	12	15	21	14	15	21	11	1	1
264	15	15-15	14	31	23	10	12	15	10	11	14	20	13	16	21	10	1	1
265	13	13-15	14	32	23	10	11	13	10	13	14	21	15	18	21	12	1	1
266	15	15-14	12	29	23	10	11	13	10	11	16	21	15	18	21	11	1	1
267	14	14-13	13	30	24	10	13	13	13	11	15	19	16	17	24	12	1	1
268	13	13-16	13	30	23	10	15	13	11	11	14	19	15	17	23	12	1	1
269	13	13-14	13	30	24	10	13	13	11	12	14	19	15	16	21	12	1	1
270	14	14-11	13	29	24	10	13	13	12	11	14	19	15	16	23	11	1	1
271	14	14-11	13	29	24	11	13	13	12	11	15	19	15	17	23	12	1	1
272	13	13-13	13	31	24	10	14	13	11	12	14	20	16	17	22	11	1	1
273	14	14-11	14	31	24	10	13	13	12	13	14	18	15	17	23	11	1	1
274	13	13-16	13	31	23	10	11	13	10	12	14	20	15	15	20	10	1	1
275	13	13-15	14	30	23	6	14	13	11	11	14	19	16	15	22	12	1	1
276	13	13-14	14	31	23	10	14	12	10	12	14	20	16	16	22	12	1	1
277	13	13-13	13	30	24	10	12	13	11	14	14	20	15	15	22	13	1	1
278	14	14-11	13	29	24	10	13	12	12	13	15	19	15	18	23	12	1	1
279	14	14-13	12	28	22	11	11	13	10	11	16	20	16	15	21	10	1	1
280	13	13-14	14	31	24	7	14	13	11	12	14	20	16	17	22	11	1	1
281	14	14-14	14	30	24	11	13	13	11	12	14	20	15	15	22	12	1	1
282	14	14-11	13	29	24	10	13	13	12	12	15	20	15	18	23	12	1	1
283	13	13-15	12	31	23	10	15	13	13	11	14	19	15	15	24	12	1	1
284	13	13-16	13	31	23	10	11	13	10	12	14	20	15	16	20	10	1	1
285	14	14-11	13	30	24	10	13	13	12	11	15	19	18	18	23	12	1	1
286	15	15-11	13	30	24	11	11	13	11	10	14	20	16	18	23	14	1	1
287	15	15-16	13	31	21	10	11	13	11	12	14	21	16	17	21	12	1	1

Valle, Cauca and Nariño, which present a similar history of peopling and are considered overall as belonging to “Mestizo” groups. One hundred and four males came from Valle, one hundred and two males came from Cauca and one hundred and two males came from Nariño. Subjects were selected among individuals who participated in paternity tests and gave their informed consent. Genomic DNA was extracted using Chelex 20%. Amplification of the 17 Y-STR loci was performed using the AmpFISTR® Yfiler™. Thermal cycling was performed using GeneAmp PCR system 2720 (Applied Biosystems). Amplifications typically contained 0.5–1.0 ng of extracted DNA. The final reaction volume used was 25 µl, and cycling was performed as described in the manufacturer's instructions (30 cycles for Yfiler system). The PCR products were typed by capillary electrophoresis using a ABI 3130 Genetic Analyzer (Applied Biosystems) according to manufacturer's instructions. Allele designation was performed according to ISFG recommendations on forensic analysis using Y-chromosome STRs using the commercial allelic ladder of the Y-Filer system¹.

2.1. Analysis of data

Haplotypes were determined by means of Excel matrix and haplotypic diversities were calculated according to Nei², using the equation: $D = (n/n - 1)(1 - \sum p_i^2)$, where n is the sample size and p_i is the allelic frequency. The haplotypes obtained in this study were compared with the haplotypes reported in the Y-STR haplotype reference database (YHRD)³. Several statistic parameters were calculated using ARLEQUIN software ver.2000: variance by AMOVA, gene frequencies and gene/haplotype diversity values.

2.2. Quality control

Correct allele calling was additionally assured by successful participation in the proficiency testing of the GEP-ISFG WG (<http://www.gep-isfg.org>) and quality testing of the <http://www.yhrd.org>.

3. Results and discussion

A total of 287 different Y-chromosome haplotypes were observed in the 308 males from three populations (Table 1). The most frequent haplotype (14-13-17-14-32-23-10-11-12-10-11-14-20-15-17.2-21-11) was observed thrice, and nineteen others only twice. Allele frequencies and gene diversity values are shown in Table 2. The highest diversity value in this study was found at locus DYS385 (0.983) followed by locus DYS458 (0.795). Haplotypic diversity found among populations in this study was 0.9977. The discrimination power, based on the number of individual haplotypes and the total number of individuals analyzed, was 93.2%. Analysis of molecular variance (AMOVA) reveals that the highest variation is mainly within populations (99.1%) in agreement with previous results in European populations⁴.

Overall, the haplotypic diversities found in these populations were higher than the ones reported in other populations^{6–13}, and equal to the one reported in a population study performed in a Valle population sample (14). This finding can be interpreted as an evidence of specific population dynamics in the Southwestern states of Colombia.

We also found allele duplications in the DYS448 STR on two individuals. These finding was confirmed through a second test of both samples. This type of variant, namely the DYS448 19-20 duplication, has not been previously reported.

We found the rare allele 10 in the DYS456 STR in one individual from Cauca, and we also detected the intermediate allele 16.3 in the DYS458 STR in a different individual from this same state.

Table 2

Allele frequencies and diversities of the 15 STR systems in the Southwestern region: Valle ($n = 104$), Cauca ($n = 102$) and Nariño ($n = 102$).

Locus	Allele	VALLE	CAUCA	NARIÑO	Southwestern region
DYS19	12	–	–	0.02	0.006
	13	0.171	0.314	0.412	0.298
	14	0.562	0.431	0.431	0.333
	15	0.181	0.147	0.098	0.113
	16	0.067	0.039	0.02	0.042
	17	0.019	0.069	0.02	0.036
GD		0.623	0.695	0.640	0.787
DYS389 I	12	0.19	0.167	0.157	0.172
	13	0.657	0.569	0.52	0.586
	14	0.152	0.245	0.314	0.236
	15	–	0.01	0.01	0.013
GD		0.514	0.594	0.612	0.573
DYS389 II	27	0.019	0.02	–	0.01
	28	0.143	0.088	0.029	0.087
	29	0.39	0.363	0.245	0.333
	30	0.286	0.265	0.431	0.327
	31	0.143	0.176	0.216	0.178
	32	0.01	0.088	0.059	0.052
	33	0.01	–	0.02	0.01
GD		0.732	0.759	0.710	0.742
DYS390	20	–	–	0.01	0.003
	21	0.07	0.069	0.029	0.058
	22	0.09	0.078	0.039	0.071
	23	0.23	0.284	0.294	0.272
	24	0.45	0.441	0.549	0.479
	25	0.14	0.118	0.069	0.11
	26	0.03	0.01	0.01	0.016
GD		0.718	0.707	0.611	0.678
DYS391	6	0.029	–	0.01	0.013
	7	–	–	0.01	0.003
	9	0.048	0.049	0.069	0.055
	10	0.514	0.686	0.569	0.589
	11	0.4	0.235	0.333	0.324
	12	0.01	0.02	0.01	0.013
	13	–	0.01	–	0.003
GD		0.578	0.476	0.566	0.547
DYS392	11	0.381	0.412	0.265	0.353
	12	0.029	0.02	0.088	0.045
	13	0.448	0.294	0.392	0.379
	14	0.114	0.147	0.137	0.133
	15	0.019	0.118	0.108	0.081
	16	0.01	0.01	0.01	0.01
GD		0.646	0.715	0.745	0.708
DYS393	8	0.01	–	–	0.003
	11	0.01	–	–	0.003
	12	0.124	0.216	0.118	0.152
	13	0.762	0.657	0.794	0.738
	14	0.086	0.108	0.069	0.087
	15	0.01	0.02	0.02	0.016
GD		0.400	0.515	0.354	0.426
DYS438	9	0.141	0.128	0.01	0.094
	10	0.23	0.282	0.294	0.269
	11	0.18	0.231	0.235	0.217
	12	0.42	0.359	0.392	0.392
	13	0.03	–	0.069	0.032
GD		0.724	0.729	0.707	0.72
DYS439	9	0.01	–	–	0.003
	10	0.08	0.078	0.098	0.087
	11	0.35	0.422	0.363	0.379
	12	0.45	0.373	0.412	0.414
	13	0.09	0.118	0.108	0.107
	14	0.03	0.01	0.01	0.016
	15	–	–	0.01	0.003
GD		0.666	0.669	0.684	0.668
DYS437	13	0.01	0.029	–	0.013
	14	0.47	0.608	0.618	0.566
	15	0.45	0.304	0.333	0.362
	16	0.08	0.059	0.039	0.058
	17	–	–	0.01	0.003
GD		0.576	0.539	0.511	0.546

Table 2 (continued)

Locus	Allele	VALLE	CAUCA	NARIÑO	Southwestern region
DYS448	17	0.001	0.01	–	0.0036
	18	0.08	0.088	0.088	0.085
	19	0.49	0.353	0.422	0.422
	19/20	–	0.01	0.01	0.004
	20	0.29	0.343	0.372	0.335
	20.4	–	0.02	–	0.007
	21	0.12	0.147	0.098	0.122
	22	0.01	0.029	0.01	0.016
	GD	0.661	0.734	0.679	0.69
DYS456	10	–	0.01	–	0.0003
	12	0.01	–	0.01	0.006
	13	0.01	0.01	0.01	0.01
	14	0.086	0.167	0.039	0.097
	15	0.438	0.451	0.5	0.463
	16	0.362	0.275	0.333	0.324
	17	0.057	0.078	0.069	0.068
	18	0.038	0.01	0.039	0.029
	GD	0.674	0.694	0.642	0.67
DYS458	13	0.01	–	0.01	0.006
	14	0.029	0.01	0.01	0.016
	15	0.143	0.127	0.216	0.162
	16	0.229	0.225	0.225	0.227
	16.2	0.01	–	–	0.003
	16.3	–	0.01	–	0.003
	17	0.257	0.314	0.333	0.301
	17.2	0.029	0.069	0.02	0.039
	18	0.21	0.186	0.176	0.191
	18.2	–	0.01	–	0.003
	19	0.048	0.039	0.01	0.032
	19.2	–	0.01	–	0.003
	20	0.029	–	–	0.01
	21	0.01	–	–	0.003
	GD	0.820	0.801	0.768	0.795
DYS635	19	0.01	–	–	0.003
	20	0.048	0.069	0.039	0.052
	21	0.21	0.196	0.206	0.207
	22	0.171	0.294	0.284	0.249
	23	0.476	0.353	0.402	0.411
	24	0.076	0.049	0.069	0.065
	25	0.01	0.039	–	0.016
GD		0.698	0.749	0.716	0.721
Y-GATA-H4	9	–	0.02	–	0.006
	10	0.029	0.039	0.039	0.036
	11	0.438	0.373	0.294	0.369
	12	0.476	0.5	0.549	0.508
	13	0.057	0.049	0.098	0.068
	14	–	0.02	0.02	0.013
GD		0.583	0.612	0.607	0.602
DYS385a/b	GD	0.9943	0.9998	0.9998	0.9979
	HD	0.9982	0.9970	0.9981	0.9977
	PM				0.000441
	CD	0.333	0.294	0.307	0.935
	UH	102	90	95	287

GD: Gene diversity, HD: haplotype diversity, PM: probability of matching, CD: chance of discrimination, UH: unique haplotypes.

The three population samples “Valle [Mestizo]”, “Cauca [Mestizo]” and “Nariño [Mestizo]” have been submitted to the Y Chromosome Haplotype Reference Database (www.yhrd.org) and were assigned to the “Eurasian/European” metapopulation (Pop-Search menu), which are defined as large pooled reference populations in the YHRD for calculating match probabilities in forensic casework⁵.

Conflict of interest statement

We, Sandra Julieta Ávila, Ignacio Briceño and Alberto Gomez declare that we have no proprietary, financial, professional or other personal interest of any nature or kind in any product or service that could be construed as influencing the position presented in the manuscript entitled: Genetic population analysis of 17 Y-chromosomal STRs in three states (Valle del Cauca, Cauca and Nariño) from Southwestern Colombia.

Funding

No funding.

Ethical approval

No ethical approval is needed.

Acknowledgements

This work was supported by Instituto Nacional de Medicina Legal y Ciencias Forenses, División de Investigación Científica, Proyecto 572, and we thank the collaborators from the Forensic Genetics group of this Institute, and Alejandro Silva for helpful discussions. This project was also registered at the Instituto de Genética Humana, Facultad de Medicina, Pontificia Universidad Javeriana, Bogotá, under No. 1889.

References

- Gusmão L, Butler JM, Carracedo A, Gill P, Kayser M, Mayr WR, et al. DNA commission of the international society of forensic genetics. Commission of the international society of forensic genetics (ISFG): an update of the recommendations on the use of Y-STRs in forensic analysis. *Forensic Sci Int* 2006;**157**(2–3):187–97.
- Nei M. *Molecular evolutionary genetics*. New York: Columbia University Press; 1987. p. 176–9.
- <http://www.yhrd.org>.
- Roewer L, Kayser M, de Knijff P, Anslinger K, Betz A, Caglià A, et al. A new method for the evaluation of matches in non-recombining genomes: application to Y-chromosomal short tandem repeat (STR) haplotypes in European males. *Forensic Sci Int* 2000;**11**:431–43.
- Willuweit S, Roewer L. Y chromosome haplotype reference database (YHRD): update. *Forensic Sci Int Genet* 2007;**1**(2):83–7.
- Gaviria AA, Ibarra AA, Palacio OD, Posada YC, Triana O, Ochoa LM, et al. Y-chromosome haplotype analysis in Antioquia (Colombia). *Forensic Sci Int* 2005;**151**:85–91.
- Kayser M, Krawczak M, Excoffier L, Dieltjes P, Corach D, Pascali V, et al. An extensive analysis of Y-chromosomal microsatellite haplotypes in globally dispersed human populations. *Am J Hum Genet* 2000;**68**:990–1018.
- Yunis JJ, Acevedo LE, Campo DS, Yunis EJ. Population data of Y-STR minimal haplotypes in a sample of Caucasian-Mestizo and African descent individuals of Colombia. *Forensic Sci Int* 2005;**151**:307–13.
- Builes JJ, Bravo ML, Gómez C, Espinal C, Aguirre D, Gómez A, et al. Y-chromosome STRs in an Antioquian (Colombia) population sample. *Forensic Sci Int* 2006;**164**:79–86.
- Builes JJ, Bravo ML, Martínez-Pancorbo M, Moreno MA, Gómez CP. Discrimination index of Y-chromosomal haplotypes in an Antioquia (Colombia) population sample. *Int Congr Ser* 2004;**1261**:275–7.
- Builes JJ, Bravo ML, Montoya A, Caraballo L, Martínez B, Moreno MA. Population genetics of eight new Y-chromosomal STR haplotypes in three Colombian populations: Antioquia, Chocó and Cartagena. *Int Congr Ser* 2004;**1261**:310–2.
- Builes JJ, Martínez B, Gomez A, Caraballo L, Espinal C, Montoya A, et al. Y chromosome STR haplotypes in the Caribbean city of Cartagena (Colombia). *Forensic Sci Int* 2007;**167**(1):62–9.
- Builes JJ, Castañeda SP, Espinal C, Aguirre D, Gómez MV, Villamarín D, et al. Analysis of 16 Y-chromosomal STRs in a Valle (Colombia) population sample. *Int Congr Ser* 2006;**1288**:219–21.